Disease Name — Propionic academia

Alternate name(s)	Propionyl-CoA carboxylase deficiency, PCC deficiency, Ketotic hyperglycinemia
Acronym	PA
Disease Classification	Organic Acid Disorder
Variants	Yes
Variant name	Late onset (> 6weeks)
Symptom onset	Neonatal
Symptoms	Episodic crises leading to neurologic damage, coma and death.
Natural history without treatment	Metabolic crises may lead to neurologic damage including mental retardation, movement disorders, seizures. coma and sudden death are also possible.
Natural history with treatment	If treatment instituted before metabolic crisis, normal IQ and development may be seen. Treatment may improve some symptoms of affected individuals.
Treatment	Protein restricted diet with supplementary medical formula, carnitine supplementation, ketone monitoring, avoidance of fasting, cornstarch supplementation, biotin supplementation. Antibiotic (metronidazole and neomycin) treatment. Human growth hormone therapy.
Other	N/A
Physical phenotype	Characteristic facies including frontal bossing, widened depressed nasal bridge, epicanthal folds, long philtrum, upturned curvature of the lips and possible hypoplastic/inverted nipples.
Inheritance	Autosomal recessive
General population incidence	1:35,000 to 1:75,000 (may be underestimate as infants may die undiagnosed)
Ethnic differences	Yes
Population	Saudi Arabia
Ethnic incidence	1:2000 to 1:5000
Enzyme location	Mitochondria
Enzyme Function	Intermediary in the metabolism of isoleucine, valine, threonine and methionine.
Missing Enzyme	Propionyl-CoA carboxylyase
Metabolite changes	Increased glycine in blood and urine, 3-hydroxypropionic acid in blood and urine, methylcitrate, tiglic acid, tiglylglycine butanone and propionyl glycine in urine.
Gene	Enzyme is made up of alpha and beta subunits coded for by different genes - PCCA and PCCB.
Gene location	PCCA = 13q32 PCCB = 3q13.3-22
DNA testing available	Not available on a routine basis, but may be available on a research basis.
DNA testing detail	No common mutations known.
Prenatal testing	Enzyme activity in amniocytes. GCMS assay in amniotic fluid. If DNA mutations known, DNA testing is possible.
MS/MS Profile	N/A
OMIM Link	www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=232000
Genetests Link	www.genetests.org
Support Group	Organic Acidemia Association www.oaanews.org Save Babies through Screening Foundation www.savebabies.org Genetic Alliance www.geneticalliance.org